DHCR24 gene

24-dehydrocholesterol reductase

Normal Function

The *DHCR24* gene provides instructions for making an enzyme called 24-dehydrocholesterol reductase. This enzyme is involved in multiple pathways that produce cholesterol. Cholesterol is a waxy, fat-like substance that can be obtained from foods that come from animals (particularly egg yolks, meat, poultry, fish, and dairy products). It can also be produced in various tissues in the body. For example, the brain cannot access the cholesterol that comes from food, so brain cells must produce their own. In one pathway, 24-dehydrocholesterol reductase is involved in the final step of cholesterol production (synthesis), converting the fat desmosterol into cholesterol. In a different pathway, 24-dehydrocholesterol reductase converts the fat lanosterol into another fat called 24,25-dihydrolanosterol. The end product of both of these pathways is cholesterol.

Cholesterol is necessary for normal embryonic development and has important functions both before and after birth. Cholesterol is an important component of cell membranes and the fatty protective covering that insulates nerves (myelin). Cholesterol also attaches (binds) to certain proteins to turn on (activate) the hedgehog signaling pathway, which is critical for normal development of many parts of the body before birth. Additionally, cholesterol plays a role in the production of certain hormones and digestive acids.

Health Conditions Related to Genetic Changes

desmosterolosis

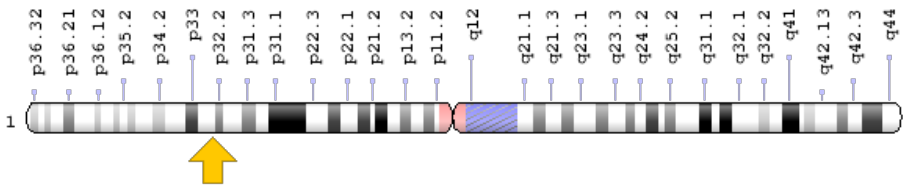
At least seven mutations in the *DHCR24* gene have been found to cause desmosterolosis. Desmosterolosis is a condition that is characterized by neurological problems, such as brain abnormalities and developmental delay, and can also include other signs and symptoms. The mutations that cause this condition change single protein building blocks (amino acids) in the 24-dehydrocholesterol reductase enzyme. As a result, enzyme activity is reduced and cholesterol production is decreased. Because the brain relies solely on cellular production for cholesterol, it is most severely affected. Without adequate cholesterol, cell membranes are not formed properly and nerve cells are not protected by myelin, leading to the death of these cells. In addition, a decrease in cholesterol production has more severe effects before birth than during other periods of development because of the rapid increase

in cell number that takes place. Disruption of normal cell formation before birth likely accounts for the additional developmental abnormalities of desmosterolosis.

Chromosomal Location

Cytogenetic Location: 1p32.3, which is the short (p) arm of chromosome 1 at position 32.3

Molecular Location: base pairs 54,849,627 to 54,887,248 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3 beta-hydroxysterol delta 24-reductase
- 3-beta-hydroxysterol delta-24-reductase
- DCE
- delta(24)-sterol reductase
- delta(24)-sterol reductase precursor
- desmosterol-to-cholesterol enzyme
- diminuto/dwarf1 homolog
- KIAA0018
- Nbla03646
- seladin-1
- seladin 1
- SELADIN1
- selective AD indicator 1

Additional Information & Resources

Educational Resources

- LIPID Metabolites and Pathways Strategy (LIPID MAPS): Desmosterol
  http://www.lipidmaps.org/data/LMSDRecord.php?LMID=LMST01010016

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DHCR24%5BTIAB%5D%29+OR+%2824-dehydrocholesterol+reductase%5BTIAB%5D%29+OR+%28seladin-1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- 24-DEHYDROCHOLESTEROL REDUCTASE
  http://omim.org/entry/606418

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DHCR24.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DHCR24%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2859
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1718
- UniProt
  http://www.uniprot.org/uniprot/Q15392

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